Archer M. (Mississippi)

Our son Archer was diagnosed with Infantile Refsum Disease (IRD) *[IRD is the term that has traditionally been used to describe the most mild presentation of Peroxisomal Biogenesis Disorder-Zellweger Spectrum Disorder (PBD-ZSD)]* in March 2012 at the age of 18 months. Our journey to diagnosis began when he was six months old and we noticed a jerking movement in his eyes. After a test called an ERG he was originally diagnosed with a congenital retinal disease called LCA. We were told he would be legally blind. Some time passed, and I remained concerned that his muscle tone was poor. His head still lagged behind when he was pulled to a sitting position, and he did not sit up until he was nearly a year old.

I happened to receive a newsletter from the Foundation for Retinal Research that suggested all LCA patients should also be tested for PEX 1 mutations. These same PEX 1 mutations had been recorded in children with Peroxisomal Biogenesis Disorder-Zellweger Spectrum Disorder (PBD-ZSD) - a fatal metabolic disorder. I immediately looked up the symptoms for PBD-ZSD and felt my heart stink. I knew Archer had this devastating disorder. The month was September, but we had to wait until December to have his PEX 1 genes analyzed. We waited three more months for the diagnosis. My mother's intuition had been right, and our lives remain forever changed.

At nearly two and a half years old, Archer has vision loss, hearing loss, and low muscle tone. He receives physical, occupational, and speech therapy. He also gets home visits with a specialist from our state school for the blind. His occupational therapist recently ordered him his very own walker, and we now are able to work daily with him at home. Archer says a few words, and can also sign "more" and "all done". He loves peekaboo, splashing in the bath, and playing with his siblings.

We are grateful for the Global Foundation for Peroxisomal Disorders (GFPD) community of families who walk this heartbreaking journey with us. We were able to attend the family conference during the summer of 2012. We were instantly bonded to these parents and children who can relate to our struggles and fears. We cried with those parents who had lost their children, and hugged those children whom we still could. Although each child is affected differently, they do share similar characteristics, and so it seemed as if Archer had just gained many more brothers and sisters. Without a doubt I knew our family had grown to include all these families affected by PBD-ZSD. We are grateful to the GFPD and to all those who support and care for our children.